Congenital scoliosis

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Summary

Congenital scoliosis is a rare disease caused by a vertebral anomaly. The defect occurs during embryological development of the spine, between the first 20 to 30 days of gestation. Other organs, such as the heart, ribs, genitourinary system or spinal cord, develop at the same time as the spine. Embryological defects may also affect these organs. The severity and the rate of deterioration of the scoliotic curve depend on the type and the site of the curvature. Thoracic volume deformity due to congenital scoliosis and rib fusion can lead to thoracic insufficiency syndrome and result in respiratory failure. Surgery is the main therapeutic option for scoliosis in an attempt to correct the curvature. There are many different types of surgical procedures, however only expansion thoracoplasty and vertical expandable prosthetic titanium ribs (VEPT) treat TIS directly.

Contents

Introduction

Scoliosis

The word scoliosis comes from Ancient Greek and means obliquity, bending. Scoliosis is defined as a curvature in the spine in the coronal plane. It is typically accompanied by a variable degree of the rotation of the spinal column. Measurement by Cobb angle (Fig. 1A) defines the scoliosis. Curves with Cobb angle less than 10* are referred to as spinal asymmetry, and have no long-term clinical significance (1).

The Cobb angle is the "gold standard" of scoliosis evaluation endorsed by the Scoliosis Research Society. It is used as the standard measurement to quantify and track the progression of scoliosis; thereby helping the doctor to determine what type of treatment is necessary. Cobb angle was first described in 1948 by Dr. John R Cobb where he outlined how to measure the angle of the spinal curve (2).
To determine the Cobb´s angle of the spine, one has to identify first the apical vertebra. The apical vertebra is the vertebra that is most displaced and rotated, with the least tilted endplates. The end vertebrae above and below are identified next. The end vertebrae are the most superior and inferior vertebrae which are least rotated and displaced, and have the maximally tilted endplate. A line is drawn along the superior end plate of the superior end vertebra, and a second line is drawn along the inferior end plate of the inferior end vertebra. The angle between these two lines is measured as the Cobb angle (3).

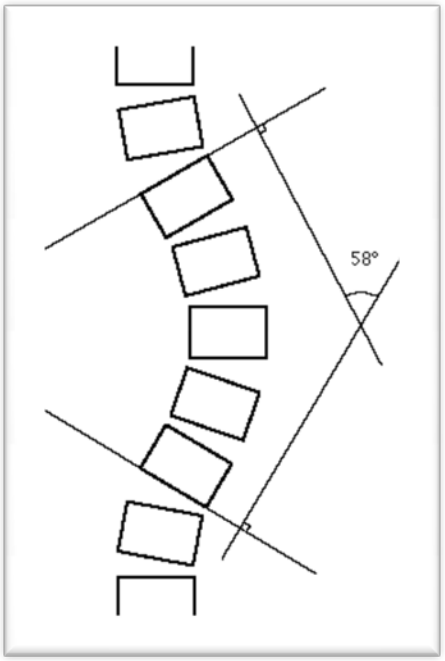


Fig. 1A

Etiologic classification of scoliosis:

- Non-structural scoliosis has no rotational component; it may be related to postural abnormalities or leg-length discrepancy. Most patients have additional symptoms related to the underlying disorder that help in making the diagnosis.
- Neuromuscular scoliosis: occurs in patients with neurologic and/or musculoskeletal disorders (cerebral palsy, muscular dystrophy…).
- Idiopathic scoliosis: is scoliosis for which there is no definite aetiology. It is a diagnosis of exclusion. We can differentiate between three subcategories:
    - Infantile: 0 to 3 years
    - Juvenile: 4 to 9 years
    - Adolescent: ≥ 10 years
- Congenital scoliosis: results from asymmetry in the vertebrae secondary to congenital anomalies. It will be further discussed below (1).

Idiopathic scoliosis is the most common form (75%), followed by neuromuscular scoliosis (15%) and congenital scoliosis (10%). (4;4)

Motivation

I chose to write about congenital scoliosis for personal reasons. A close family member of mine has this disease and I realised that I knew too little about this illness. Orthopaedics is not a major subject during the medical syllabus at the University of Oslo. It is barely mentioned in the semester during which we learn about the muscular- and skeletal system. Pathology in muscles and bones is not so much in focus because of the lack of time. Regular anatomy should logically be of priority. Scoliosis is not only unknown to many, but it is also a rare disease. In my opinion it is important to know a little about it because the therapeutic success is excellent when diagnosed and treated at an early age. Usually it is detected at birth, but there are cases where it is discovered at later age by surprise. These cases seldom occur, but may occur. I am very glad I wrote about congenital scoliosis. I found it surprisingly interesting and eye-opening.

Methods

The literature was systematically searched in Google Scholar, PubMed, the American Journal of Bone and Joint Surgery, NEL, and UpToDate from the earliest date of each database, with special focus on well-known international orthopaedic surgeons specialised in scoliosis.

Congenital Scoliosis

Congenital scoliosis is a lateral curve of the spine that is due to the presence of vertebral anomalies that cause an imbalance in the longitudinal growth of the spine. These vertebral anomalies occur in the first six weeks of intrauterine life, when the anatomical pattern of the spine forms from the mesenchyme. The vertebral abnormality is present at birth, but it may not become evident until later in childhood (5).

These anomalies may cause a wide spectrum of deformities that affect the balance of the developing spine. These deformities may be mild, causing subclinical curvatures, or severe, causing early, crippling, progressive scoliosis often associated with neurologic complications, pulmonary restriction, cor pulmonale, and premature death (6).

Incidence

Congenital scoliosis occurs more often in girls than boys, with a ratio of 2.5 to 1 (6), and has an incidence of approximately 0.5 to 1/ 1000 births (7).
Most severe curves are clinically evident during the first years of life or between the ages of nine and thirteen (the periods of most rapid growth of the spine) (5).

Classification

The current classification system is based on the radiographic appearance of the vertebral malformation. We can differentiate between two main categories: the type of anomaly and the site of the apex (the area of greatest displacement from the midline of the body).

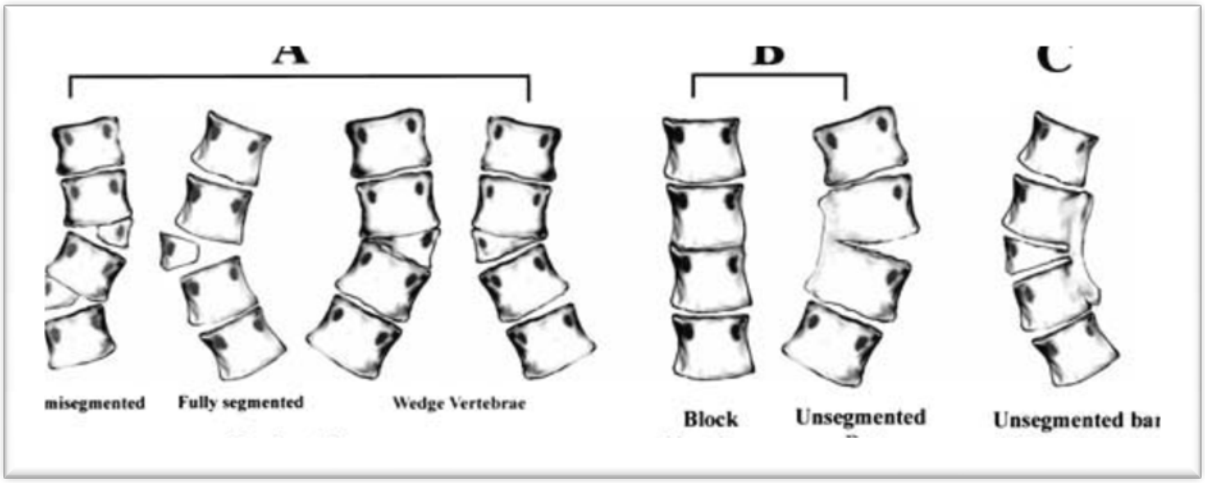
Type of anomaly:

- Simple anomalies:
    1) Failure of segmentation: portions of adjacent vertebral elements fail to divide. These errors can produce:
        - Unilateral unsegmented bar: does not contain growth plates and can therefore not grow longitudinally. At the same time, normal or nearly normal growth can occur on the opposite site. The longer the unsegmented bar is, the larger the curve in a specific region.
        - Unilateral unsegmented bar with contralateral hemivertebrae at the same level: is the type of scoliosis with the worst prognosis. It may be difficult to see the hemivertabrae radiographically at an early age since it gets obscured by of the severity of the curve.
        - Bilateral failure of segmentation or block vertebrae (block vertebrae): is due to a bilateral failure of segmentation, its longitudinal growth is impaired on both sides of the spine, but not always asymmetrically.

    2) Failure of formation of a vertebral body: produces:

- Hemivertebra (complete): acts as an enlarging wedge on the affected side of the spine. It is never as severe as in those with a unilateral unsegmented bar. Not all hemivertebrae produce the same level of imbalance.
- Wedge vertebra (partial): is due to a unilateral partial failure of formation of a vertebra with retarded longitudinal growth.

- Complex anomalies that include both segmentation and formation errors (5).



Diagrammatic representation of classification system of congenital scoliosis:
*(**A**) Failures of formation; various types of hemivertebrae (semisegmented, fully segmented, wedge vertebra).*
*(**B**) Failures of segmentation; block vertebra, unsegmented bar.*
*(**C**) Mixed; combination of hemivertebra and unsegmented bar. (8).*

Site of the apex: often coincided with the site of the anomaly.
- Upper thoracic curve: the apex lies between Th2 and Th6.
- Lower thoracic curve: apex between Th12 and L1.
- Thoracocolumbar curve: apex between L2 and L4.
- Lumbosacral curve: apex at L5 (5).

Among vertebral anomalies causing scoliosis, unilateral unsegmented bar is the most form for anomaly, closely followed by hemivertebrae, complex anomalies, a unilateral unsegmented bar with contralateral hemivertebrae and block vertebrae. The least common is a wedge vertebra.
The most common site for the congenital scoliosis is the lower thoracic region, followed by the upper thoracic, the thoracolumbar and lumbar regions. The least common site is the lower sacral region (5).

The malformations may occur on either the right or left side of the sagittal plane of the body resulting in “pure” scoliosis; or in the anterior or posterior of the coronal plane resulting in “pure” kyphosis or lordosis. Combined deformities are most common, producing scoliosis and sagittal plane deformity (8).

Natural History

The severity of the congenital deformity and the rate of deterioration depend on both the type of anomaly and the site of the anomaly. The rate of deterioration is not constant; it tends to increase at the age of 1-5 years and at pubertal growth (5).

In order to understand the variable prognosis of congenital scoliosis, it is necessary to associate the principles of normal growth of the spine with the pathological anatomy of the various types of congenital vertebral anomalies. Normally, longitudinal growth of the spine results from the sum of the total growth occurring at the end-plates on the upper and lower surfaces of the vertebral bodies. This occurs equally on either side of the spine, so that the spine remains straight without scoliosis. A congenital vertebral anomaly can cause a growth imbalance due to

a deficiency in either the number of end-plates or their rate of growth on one side of the spine. The resulting lateral curve is of a severity proportional to the degree of the growth imbalance (5).

The growth imbalance caused by vertebral anomalies is the most important feature in classifying a congenital scoliosis with regard to its prognosis. The rate of deterioration is worse if the curve is present before the age of ten years because of the adolescent growth spurt (5).

The most progressive type of scoliosis is that due to a unilateral unsegmented bar with a contralateral hemivertebra at the same level. This type of curve can progress at a rate of 10 to 12 degrees per year. Scoliosis caused by an isolated unsegmented bar is the next most severely progressive type of curve, followed by scoliosis by two consecutive free hemivertebrae on the convex side. Progression of a cure caused by a single hemivertebra is difficult to predict; this type of curve may progress severely, slowly or not at all.
It is important to know whether a curve is progressing. This determination depends on accurate and precise monitoring, both clinically and radio graphically (9). Regarding the site of the apex, the sites with the worst prognosis were the lower thoracic and the thoracolumbar curves (5).

There are some *secondary features* that are significant to the over-all disability and deformity of the patient. Upper thoracic curves usually produce a cosmetic deformity due to an elevation of the shoulder or tilting of the head (less frequent) (5).

Patient with upper thoracic curves with the apex at the levels of Th5, Th6 and Th7 often develop a secondary structural curve in the lower thoracic or thoracolumbar region. Initially it is compensatory and correctable, but later it becomes fixed and tends to deteriorate faster than the primary, congenital, curve. The lower secondary curve has a tendency to rotate more. The large rib hump usually constitutes a major deformity.

Patients with a lower thoracic, thoracolumbar or lumbar curve seldom develop a secondary curve to balance the congenital curve because there are too few normal mobile vertebrae between the anomaly and either the upper end of the spine or the sacrum. What these patients usually develop is some degree of pelvic obliquity and an apparent shortening of one of the lower limb. Decompensation or listing of the upper part of the body to one side is a common finding associated with a more severe lower thoracic and thoracolumbar curves. It could result in a very severe deformity (5).

Thoracolumbar curves can be caused by a single hemivertebrae at the lumbosacral junction. The hemivertebra can force the lumbal spine to take off obliquely from the sacrum. In an attempt to balance the body, a secondary lumbar or thoracolumbar curve can develop. But this may not be enough to prevent the trunk from listing to the opposite side that of the hemivertebra (5).

Ideally, congenital scoliosis should be diagnosed while the patient is still young and the curve small. At that point, a curve that is at risk for progression can be recognized and a appropriate prophylactic course of surgical treatment can be planned in order to prevent a severe deformity (5).

Formation of the spine

The spine is formed during a process called somitogenesis during the first 20 to 30 days of gestation, the third and fourth week of gestation. In this process, segments of mesodermal tissue called somites, are formed in pairs surrounding what will eventually become the spinal cord. These somites are regularly sized and spaced, and this careful organization is essential for the normal patterning of the spine. If somitogenesis is disrupted even slightly, as has been done in animal models, congenital vertebral defects similar to those in congenital scoliosis have resulted.
The somites mature producing sclerotomal cells that migrate and surround the developing spinal cord to form the bones of the vertebrae. These same somites also form the axial muscles that connect the vertebral segments, and the ribs associated with the thoracic vertebrae. Many other organs and tissues are being made during this important time in development, including the heart, kidneys, brain and limbs.
Developmental studies in animal models show that a series of developmental genes regulate somitogenesis and when these genes are disrupted, vertebral deformities can result. The genes in the notch-gene family for instance, regulate development of vertebral precursors during the embryogenesis. Other genes associated with congenital scoliosis are: Pax1, DLL3,… (10).
Environmental factors, such as hypoxia caused by carbon monoxide, can also disrupt somitogenesis and cause congenital vertebral anomalies. The incidence and severity of these are directly related to dose and time of

exposure (8). Maternal diabetes and ingestions of anti-epileptic drugs during pregnancy also have an adverse effect (10).

Associated Anomalies

Anomalies occur at other sites than the spine in 30% to 60% of children with congenital spinal anomalies. Embryologic development of the spinal cord coincides with the development of other organ systems. The most common sites are the genitourinary tract, the cardiac system, the spinal cord and the cervical spine (6).

The most frequently associated defect is found in the ***genitourinary system***. 20% to 40% of patients with congenital scoliosis have renal anomalies (6). Most patients have no complaints and show no symptoms that suggest that they might have a possible urologic disorder. All the same, it is advisable to do an ultrasound of the kidneys of all children with congenital scoliosis, regardless of the severity and location of the spinal deformity. Unilateral renal agenesis is the most common urologic abnormality. A solitary kidney is susceptible to infection, obstruction, stone formation and renal trauma. Other prevalent renal malformations are a duplication of the kidney pelvis or ureter or both; obstructive uropathy, renal ectopia and horseshoe kidney (11).

About 10–15% of patients with congenital scoliosis have ***congenital heart defects***, such as atrial or ventricular septal defects, tetralogy of Fallot or transposition of the great vessels (12). The abnormalities can be detected during the routine preoperative appointment, or the patient may have a long cardiac history. Patients undergoing an operation for a congenital spine deformity have to undergo a screening echocardiogram, with a referral to a cardiologist if needed (13).

In approximately 20% of patients with congenital scoliosis, ***spinal cord developmental defects*** have been observed, including tethered cord, fibrous dural bands, diastemetomyelia, or intradural lipoma (8). Studies have shown that patients with congenital vertebra anomalies have a significant smaller spinal cord than healthy children at the same age (14). These spinal cord abnormalities are frequently associated with cutaneous changes (hair patches, dimples, pigmentation) and various abnormalities of the lower extremities, including flat feet, cavus feet, vertical tali, clubfeet, or asymmetric reflexes (8). The absence of neurological deficit does not rule out intraspinal dysraphism (13).

Other defects associated to congenital scoliosis are ***absent or fused ribs***, since the ribs derive from the same embryonic origins as the vertebrae (8). It is frequently associated with thoracic and thoracolumbar scoliosis probably due to a unilateral failure of vertebral segmentation. Rib anomalies can be divided into two categories, simple and complex. A patient with a simple rib anomaly has either a localised fusion of two or three ribs, or a small chest wall defect. The most common type of anomaly is the rib fusion. The most common complex rib anomaly is the extensive fusion of multiple simple ribs associated with a large adjacent chest wall defect due to an absence or deviation of ribs.

Extensive rib fusions affecting the hemithorax on the concavity of a congenital scoliosis can in growing children act as a powerful lateral tether to further imbalance the growth of the spine, which is already being deformed by asymmetrical vertebral growth. An extensive thoracic congenital scoliosis associated with fused ribs may affect thoracic function and the growth of the lungs in young children and lead to thoracic insufficiency syndrome. An imbalance in the mechanical thrust of the ribs can also adversely affect spinal growth as well the function of trunk muscles and the pressure within the thorax (15).

Sprengel`s shoulder, also known as ***congenital elevation of the scapula***, is an shoulder anomaly due to abnormal descent, altered position and anatomy of the scapula (16). It is commonly associated with patients with cervicothoracic or thoracic scoliotic deformities (15).

Other anomalies that can be expected are: esophageal atresia, tracheoesophageal fistula, diastematomyelia, anal atresia, facial asymmetry, bladder and cloacal extrophy.
Syndromes including congenital vertebral defects are:

- Klippel-Feil syndrome (short neck, low posterior hairline, fusion of cervical vertebra)
- Goldenhar`s syndrome (associated with craniofacial anomalies like microtia and epibulbar dermoids due to abnormal branchial arch development)
- Incontinentia pigmenti (hyperpigmented wholrs and streaks associated with eyes, skin, hair, nail, teeth and central nervous system anomalies
- VACTERL (Vertebral malformations, Anal atresia, Cardiac malformations, Tracheoesophageal fistula, renal and Radial anomalies and Limb defects) associations (7).

Clinical consequences of scoliosis

As the spinal curvature progresses, pulmonary function can be compromised if the deformity occurs in the thoracic region (7). Progressive scoliosis alters lung function by reducing the compliance of the chest wall and rotating intrathoracic contents, producing an increasingly asymmetric lung size (17). Over time, the scoliotic disorder changes its nature: from a mainly orthopedic issue, it becomes a severe pediatric, systematic disorder with TIS, cor pulmonale, and hypotrophy. In the most severe cases, these alterations can be fatal (18).

Cardiorespiratory failure usually develops in patients with a Cobb angle greater than 90 degrees. It is more likely to occur in those patients with a thoracic curve, an associated thoracic lordosis, structural vertebral anomalies such as congenital scoliosis, or respiratory muscle failure. Severe scoliosis leads to respiratory failure due to alveolar hypoventilation and circulatory failure because of arterial hypertension and cor pulmonale (19).

Thoracic volume deformity in congenital scoliosis should be carefully identified radiographically and by CT-scans, and classified into the different thoracic deformity categories. This will help to understand what surgical treatment is most adequate for the patient.
We can differentiate between three different categories of thoracic deformities:

- *Type I*: is due to absent ribs and scoliosis. The flail lung prolapses into the chest with inspiration leaving minimal space for the lung to expand during expiration. The lung is usually smaller than normal. Patient with massive rib-defects tend to have a high mortality rate in infancy because of respiratory failure.
- *Type II*: is the most common type of deformity. It is due to fused ribs and scoliosis. The concave hemi-thorax constricts the underlying lung at birth. As the deformity progresses due to both the increasing scoliosis and the loss of relative lung growth from the rib constriction, the deformity aggravates. Figures 2A, 2B, and 2C show a classic example of this deformity.
- *Type III*: is a lethal deformity, either from the thorax being too short (type IIIA) or too narrow (type IIIB). It has no relationship to scoliosis. Affected patients develop a severe extrinsic, restrictive lung disease because the significant loss of lung volume and the stiffening of the chest wall (20).

Fig 2A.

2B. 

2D. 

Figures showing a type II Thoracic Lung Deformity with fused ribs and scoliosis from an anterior (A), posterior (B) and lateral (C) view.

Extensive thoracic congenital scoliosis associated with fused ribs may affect thoracic function and growth, and have an adverse effect on the function and growth of the lungs, leading to thoracic insufficiency syndrome (21).

Thoracic insufficiency syndrome (TIS) is defined as the inability of the thorax to support normal respiration and lung growth.(20). It was firstly described by Dr. Robert Campbell in 2004.

Primary thoracic deformity may result from fused ribs (limiting lung growth of the underlying lung) or absent ribs (which results in thoracic instability with compromise of the respiration). Secondary deformities of the rib cage result from curvature, rotation and foreshortening of the thoracic spine (21).

The rib cage of the concave side of the curve can be described as the concave hemithorax, the chest wall on the contralateral side, the convex hemithorax. The portions of the chest wall involved in the rib fusion can be considered areas of segmental hypoplasia of the hemithorax (21). In cases of extensive rib fusion, the concave hemithorax with limited growth potentials serves as a powerful lateral tether to spinal growth, by increasing the unbalanced growth of a spine that is already deformed by asymmetric vertebral growth (21).

Growth of the normal lung and thoracic cage is parallel to one another after birth. The volumes of both structures increase in a non-linear fashion over the first two decades of life, with rapid growth occurring before 3 years of age and again during the pubertal growth spurt (18). Anatomical boundaries of the thorax limit lung growth. Any spine or rib cage malformation reducing the thoracic volume early in life will adversely affect the size of the lungs at skeletal maturity. Thoracic anomalies may help explaining why patient with congenital scoliosis have a lower vital capacity on pulmonary function testing than patients with idiopathic scoliosis (21). There is no significant correlation between the Cobb angle and the pulmonary function (22), but it is known that children with scoliosis in an early age have poorer pulmonary test results than those with a later debut of the scoliosis(17). Agenesis or aplasia can be seen. The alveoli fail to multiply and may even atrophy in a developing compressed lung as a result of severe scoliosis during childhood (22). The distance between T1 and T12 has to be greater than 22 cm to avoid development of TIS. When it is under 18 cm is it critical.

Respiratory function measurements

Respiratory function should be repeatedly assessed in patients with scoliosis in order to detect a developing thoracic insufficiency syndrome. The history of respiratory symptoms, the findings of physical examination, radiographs, CT-scans and routine measurements of the lung function (including: static lung volumes, forced spirometry, lung perfusion scans, blood gasses and respiratory muscle strength) and CVC are the main assessments. The patient has to be 5 years or older to undergo the function testing (19).

Standard lung function test show the loss of vital capacity of the lungs reflecting a restrictive pattern of lung function. These tests reflect many conditions: the primary loss of lung tissue, the additive effects of reactive airway disease, and the secondary loss (because of volume- and function-reduction of the thorax) (21).
There are many sources of error in the standard lung function tests that have to be considered. These procedures depend on the cooperation ability of the patient. Patient under five years of age cooperate poorly. The vital capacity measures depend on the height, but this can be misleading with regards to patients with scoliosis since their spine is often shortened because of the vertebral malformation. Blood gas analyses can be inaccurate when a child is hyperventilating from crying (21).

Haemoglobine can be a good objective surrogate measure for lung function because it is independent of cooperation. Studies among people living in high altitudes show that there is an association between haemoglobine and chronic hypoxemia. A Complete Blood Count (CBC) is a good method for screening children for TIS and evaluating the outcome of the surgery (23).

Consequences of TIS
The prevalence of thoracic insufficiency syndrome is unknown. Many children with thoracic and spinal deformity may walk around undiagnosed with a latent thoracic insufficiency syndrome. When a thorax with fused ribs cannot support normal respiration, the child may compensate with hyperventilation and reduction of activity level. The child will appear clinically normal, while the lungs may be compressed with development of atelectasis and the expansion of the lungs for respiration becomes dependent of the diaphragm (21).

Patients can tolerate this condition clinically for a long time, but sooner or later they will be depend on oxygen or ventilation support. By that time, it will be impossible to correct the problem. It is thought that the lungs might stop growing at the age of eight years. Any form for treatment of the spine and the rib-cage deformity before that time, may enable the growth of all the components of the thorax. This can in return stimulate the growth of the lungs, so that they get the size needed in adulthood (21).

The thoracic deformity and function are directly proportional, when the first one progresses, so does the latter. A decrease in the percentage of vital capacity, an increase of respiratory rate or increasing clinical respiratory symptoms such as dyspnoea, are signs of progressive thoracic insufficiency (21).

Effects of TIS on nutrition
Another factor to consider in patients with TIS is the prevalence of malnutrition. Children with pulmonary insufficiency characteristically have poor nutrition as the energy expenditure from the extra work of breathing outweighs the nutritional gain of eating. Some children lack of sufficient pulmonary reserves to hold their breath long enough to swallow while eating. This can contribute to the nutritional depletion. Nutritional consultation is an essential part of the pulmonary care of TIS patients. For growth and development, the calorie intake must be greater than the energy demands. BMI cannot be used as a measure for the nutritional status because it depends on the height (24).

Treatment

The therapeutic options in cases of congenital scoliosis include conservative or surgical approaches. The goal of the treatment is to prevent development of a severe malformation. Early diagnosis, anticipation and prevention of deterioration are the vital actions.

Congenital scoliosis is a challenging condition, both for the patient and his or hers family; it may seriously impair their quality of life for a long interval of time. Any form for treatment requires good compliance from the patient and also caregivers. Surgical treatment often requires repetitive surgery over a period of time. For this reason it is essential to build an understanding and supportive relationship with the patient's family, in order to avoid concerns and conflicts about the clinical situation of the child (18).

Bracing

Most congenital curves are nonflexible and therefore resistant to correction by bracing. Bracing is a good measure in order to prevent the progression of secondary curves that can develop above or below the congenital curve causing imbalance (10). The Milwaukee brace is the best brace choice for upper curves. A TLCO (thoracic/lumbar/sacral orthosis) can be used in lumbar curves but is mostly used only in the USA.
The treatment should last until skeletal maturity is reached. The brace will not correct or arrest the possible development of the curve, but it may delay the progression and maintain flexibility, allowing surgery at a later stage. Bracing has no effect on short, sharp, rigid curves (6).

Surgery

The main goal of surgery is to balance the spine, provide some correction of deformity and arrest curve progression (if possible). The surgical options are numerous and depend on the type of anomaly, the degree of deformity and the age of the patient. Early diagnosis and surgical treatment before severe deformity develops is the main goal (25).

Neurological damage during surgery occurs more frequently during correction of congenital scoliosis than in other type of scoliosis. Intraoperative neuromonitoring for the detection and possible prevention of neurologic injury during spinal procedures is of great importance (26).

- ***Spinal fusion***

Very little is known about the long-term effects of early spinal fusion done in patients under 8 years of age. Recent studies have shown the prevalence of life-threatening side effects from early spinal fusion. These events have triggered a search for other methods (listed below) to treat a spinal deformity without the need for spinal fusion at an early age. Spinal fusion is used as a more terminal form for treatment, when the newer methods have corrected as much as possible of the curvature.

The main obstacle for the new methods is the inability to come up with clear indications and contraindications for them due to the low number of treatable candidates to treat. One has to be cautious when evaluating these results (18).

- ***Convex epiphysiodesis***

Convex epiphysiodesis prevents future deformity and requires growth to obtain correction over time. The ideal anomaly is a unilateral failure of formation without any associated deformity. An anterior and posterior fusion on the convex side of the curvature is done by removing a lateral half of adjacent discs. The concave side retains its growth potentials and allows for some correction as the child grows (10).

- ***Hemivertebra excision***

The ideal candidate for a hemivertebra excision is a patient younger than five years of age with a fully segmented hemivertebra at the junctional regions of the spine (cervico-thoracic, thoracolumbar or lumbosacral) (10). During this type of surgery, an incision is made anteriorly (though the chest or abdomen), posteriorly (along the back) or at both places. The most common access is the posterior approach. Once the hemivertebra has been exposed and removed, metals screws are placed into the vertebra above and below the hemivertebra site, and attached to metal rods. These rods are attached in order to stabilize the vertebrae above and below the hemivertebra. An implant is used to secure correction and bony healing. Before closing up, X-rays of the spine are taken to confirm the correct localisation of the screws and rods. Possible complications of this procedure include infection, bleeding, nerve and spinal cord injury (27).

- ***Correction with instrumentation and fusion***

The partial or complete correction of the deformity depends on the type, site and degree of the curvature (10). Spinal fusion is a procedure in which the abnormally curve is straightened and the individual bones of the spine are fused into one long straighter bone. Screws and rods are inserted to hold the spine straight while the fusion heals. It is usually performed when the curve reaches a point where non-operative measures are unable to prevent further progression. In this surgery, an incision is made exposing the spine. The approach can again be anterior, posterior (most frequently used) or anterior/posterior. Once the spine is exposed, metal screws are placed into the bones of the spine involved in the curve. The surgeon places the rods into the screws to correct and straighten up the spine. Once the spine is straightened, a bone graft is placed to help the bones in the spine to fuse, and the incision is closed up (27).

- ***Expansion thoracoplasty and vertical expandable prosthetic titanium ribs (VPTR)***

The main goal of VEPTR implants is to treat a thoracic insufficiency syndrome (TIS) and a congenital spinal deformity. TIS is defined as the inability of the thorax to support lung growth, respiratory function or spine development due to congenital and acquired chest wall, spine and other syndromic deformities. Conventional spinal arthrodesis for progressive congenital spinal deformity may the thoracic insufficiency worse by stopping growth of an already short spine.

Campbell and co-workers introduced the use of expansion thoracoplasty and VEPTR chest wall distraction to treat both the spine and the chest wall. The first procedure consists of one or more opening wedge thoracostomies and insertion of one or more VEPTR devices. The thoracostomies of the hemithorax are

expanded maximally at the initial procedure and the devices are placed to stabilize the correction. Repeated surgeries at appropriate intervals are performed in order to lengthen the VEPTR implants and further correct the deformities in the thorax and spine. Expansion thoracostomy and the use of VEPTR devices directly treat segmental hemithoracic hypoplasia by lengthening and expanding the constricted hemithorax. It also treats scoliosis indirectly without the need for spine fusion and with a probable benefit for the underlying lung (28). Increased volume of the constricted hemithorax and total lung volumes obtained with the surgery are usually maintained at the follow up (29).

Indications for VPTR

One of the surgical indications for thoracostomy and VEPTR implants is the combination of rib fusion and progressive scoliosis. When the fused ribs are extensive or when conventional treatment requires an extensive thoracic spinal arthrodesis, then VEPTR may be a logical choice if fused ribs or constricted hemithorax are present (29). Patients with this combination of diseases have a severely restricted thorax that does not allow the lungs to develop properly. Studies show that patients with a widespread thoracic scoliosis treated with rib fusion had a significant reduction in pulmonary vital capacity values later on. Other indications are hypoplastic thorax syndrome or flail chest (28).

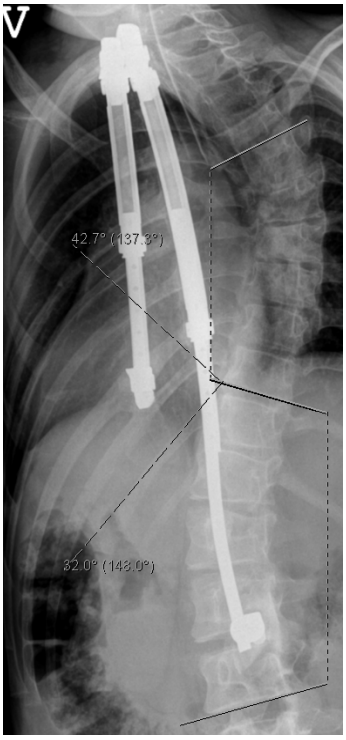
The titanium rib implant

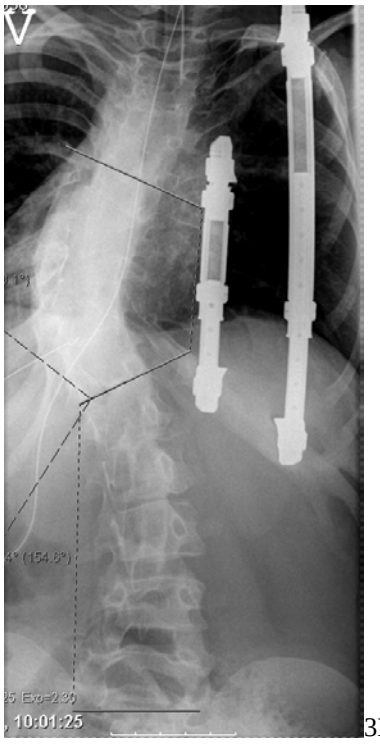
The titanium rib is an implantable, expandable prosthetic device used together with opening wedge thoracostomy to accomplish an expansion thoracoplasty. The Vertical Expansion Prosthetic Titanium Rib (VEPTR) implant is a sliding titanium construct, roughly rectangular in cross section with two standard curvatures conformed to the shape of the thoracic cage into which it is being implanted. The construct is implanted vertically along the chest wall by attaching each end to healthy ribs above and below the site of abnormality. It not only distracts and expands the chest wall, but it also acts as a protective splint over areas where ribs are missing and can indirectly apply distraction to the spinal column, controlling or improving spinal deformity (28).

The superior and inferior sections of this device consist of two pieces each, a cradle and a cradle end half. The semi-circular end of the cradle can be adjusted to different angles to accommodate patient anatomy. It is connected to the cradle end half by a cradle lock to encase the rib. The cross section of the proximal ends of the rib cradles is “T-shaped” for enhanced strength. The superior and inferior end cradles attach to the rib sleeve by distraction locks. The rib sleeve is the central section of the construct. It serves as a track into which the cradles slide. The hole in the rib sleeve lines up with one of the blind holes on the rib cradle. The position of the inferior cradle assembly along the rib sleeve depends on the desired length of the overall rib prosthesis construct.

Implantation and assembly

The superior cradle is placed over the patient`s upper ribs and secured together with a cradle lock, encasing the ribs. The rib sleeve slides onto the proximal end of the superior cradle and is secured by a distraction lock. The inferior cradle slides into the rib sleeve and is positioned and secured on the lower healthy ribs. The inferior cradle is distracted to the desired length, and the rob sleeve and inferior cradle are secured together with a distraction lock. When the scoliosis extends to the lumbar area, a lumbar extension can be used in place of the inferior rib cradle and cradle end half. The distal part of the lumbar extension is a 0,6 mm straight rod that allows for attachment to the lumbar spine with a low profile spinal hook (28). Figures 3A and 3B (below) show two implanted VEPTR devices.



3A.


3B.

Figure 3A: Medial pole from the fifth rib to the fused caudal ribs. Lateral pole from TH2-level to the same caudal fixation point.
Figure 3B: Typical VEPTR location: medial pole, from the second and third rib, with a supra-laminar hook at L3-level. Lateral pole from the second and third rib to the ninth rib.

Expansion Thoracoplasty Techniques

Enlargement and lengthening of the constricted hemithorax and indirect control of spinal curvature is achieved by one or multiple opening wedge thoracostomies. If a congenital chest wall defect is present, the remaining soft tissues may be incised in line with the ribs, in order to allow an opening wedge. If there are bony fusions of ribs, fused ribs are separated with an oscillating saw, protecting the underlying pleura, and opening wedge thoracostomy produced in the newly created interval between previously fused ribs. When there are multiple contiguous fused ribs, ribs may be divided in groups and opening wedge thoracostomies are performed between groups (28).

Posteriorly the thoracostomy must extend back to the transverse processes to permit expansion between adjacent ribs. Confluent bony jars joining ribs medial to transverse processes should be resected down to the vertebral column. Anteriorly, the thoracostomy must extend to near the costal cartilage to permit free expansion of the thoracostomy interval (28).

The thoracostomy is spread slowly to allow the intact parietal pleaura to stretch. If the pleaura is torn, artificial pleura of Gore-tex sheeting is placed loosely over the underlying lung and sutured at the periphery of the thoracostomy to parietal pleura.

Once the desired correction of the hemithorax is achieved, the assembled VEPTR devices are implanted unexpanded, then tensioned 0,5 cm to stabilize it. Excessive initial distraction may lead to premature cut out of the device, but correction and expansion are easiest to achieve at the initial procedure(28).

Patients are usually hospitalized for 7-10 days. Intubation and ventilator support are only required during the operation. Children stay in the Intensive-care unit (ICU) for 2 days postoperative before they return to the ward.

Device lengthening and Exchanges

Elective lengthening of the implanted devices are usually planned at 4-6 months intervals, depending on the severity of the disease, the stiffness of the spine, the age and prior surgeries. If there are any complications, such as intercurrent illnesses or problems travelling to the hospital, the lengthening intervals are more widely spaced.

Contraindications

There are some contraindications for the use of VEPTR devices (30):

- Insufficient bone strength in the ribs or spine where the device would attach.

- Ribs are absent either posterior or anterior to where the device would be attached.
- Non-functioning diaphragm.
- Insufficient tissue to cover the VEPTR device.
- The patient is younger than six months of age.
- Skeletal maturity is reached (14 years for girls, sixteen for boys).
- Known allergy to device material.
- Infection near or at the surgical site.

Complications

Some possible complications of this surgery are (30):

- Failure of the device to achieve stabilization or support the thoracic region.
- Migration on the spine due to fracturing or disassembly of the device.
- Development of an allergy to the implanted device.
- Development of an infection at the surgical site or within the respiratory system.
- Neurological complications occur more frequently in VEPTR- procedures than in other types of spinal surgeries. The most common injuries are brachial plexopathy and spinal cord damage (26).

Nutritional status in VEPTR children

Studies show a significant improvement in the nutritional status of children after a VEPTR operation. A possible explanation for this is that the parents and health care providers pay better attention to the child in the postoperative period, resulting in a better diet and weight gain independent of the VEPTR device. However, this gain appears to go on many months after the surgery, when the period during which the child receives more attention is over. Adequate nutrition is essential for normal wound healing and reduction of postoperative complications. Nutrition is especially important for VEPTR surgery, because up to 4 devices may need soft tissue coverage, and repeated lengthening occurs once or twice a year for a long time (24).

Life quality among VEPTR children

The main goal of expansion thoracostomy and VEPTR implantation is to improve the quality of life of the child with congenital scoliosis and to decrease the burden of his or hers caregivers. Children with TIS have lower physical domain scores and higher caregiver burden scores, but similar psychosocial domains than healthy children. Children with TIS show a lower quality of life in physical domains before VEPTR implantation (than healthy children) because they often experience limitations in daily activities, like for instance playing with friends, for example. Poor lung function and deficient breathing mechanisms holds them back. They also often suffer from significant comorbidities that directly impact their physical health. There is a direct correlation between the health of a child and the physical and emotional impact on their parents. The child`s health has a considerable impact on family relationships. Parents experience a great amount of distress and limitation of their personal time, because of their child`s physical health, emotional well-being, attention or learning abilities, his or hers ability to get along with others and general behavior. It requires a great amount of resources to take care of a child with TIS, including long-term medical services and emotional support. The family unit is a decisive aspect of the child`s health. There is no significant short-term improvement in the quality of life of children and burden experienced by their parents after VEPTR implantation. The results may change in the future as the treatment may show its efficacy when avoiding worsening of the child`s health status and reducing the high mortality risk associated with untreated TIS (31).

The effect of VEPTR

The best results on respiratory functions were found among patients that have been operated at an age of two years or younger. This is the time when there is more rapid lung growth by lung alveolar cell multiplication. The predicted normal vital capacity is higher among patients from this group. The curves decreased immediately postoperatively and after multiple lengthening of the device. Even the rigid portion of the curve can show improvement (32). The children even show a nearly normal growth compared with children at the same age (18). Once skeletal maturity is reached, some patients will require a spinal fusion. Conducting a spinal fusion on a young child may negatively impact the potential skeletal growth of the child and the fusion will not provide any support for the chest wall (30).

Conclusion

Severe congenital scoliosis is a disease with a difficult approach. It is not easy to decide which therapeutic option is the best for the patient, because he or she often presents a great variety of symptoms alongside to the existence of a spinal curvature. The most important thing to start with is to refer the patient to an orthopaedic surgeon specialised in scoliosis. It is a very rare disease, and very few surgeons have enough experience on managing

and operating on such patients. Before any therapeutic option is discussed, the patient undergoes an extensive clinical examination. Special attention is paid to organs which usually present anomalies related to the scoliosis. These are the kidneys, the lungs, the heart, and the spinal nerves. The lung function is often impaired in patients with congenital scoliosis and fused ribs. They often develop Thoracic insufficiency syndrome (TIS), which later on can end up in cor pulmonale and death if not treated early enough. Expansion thoracoplasty and VEPTR-implants have shown to treat TIS directly and the scoliotic curve indirectly by enlarging and lengthening the constricted hemithorax. The curves decrease almost immediately postoperatively and even more after multiple lengthening of the VEOTR-device. Even the rigid portion of the curve can show improvement

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